

DIAGNOSTIC METHODS FOR POMPE DISEASE AND OTHER LYSOSOMAL STORAGE DISEASES

Abstract

5 Provided are methods of screening subjects for lysosomal storage
diseases, preferably glycogen storage diseases, using a tetrasaccharide as a
biomarker. In a more preferred embodiment, subjects are screened for
Pompe disease (*i.e.*, glycogen storage disease type II). Also provided are
neonatal screening assays. The present invention further provides methods
10 of monitoring the clinical condition and efficacy of therapeutic treatment in
affected subjects. Further provided are methods of measuring a
tetrasaccharide biomarker by tandem mass spectrometry, preferably, as part
of a neonatal screening assay for Pompe disease.